In-Depth

Overview

Points To Remember About Marfan Syndrome

- Marfan syndrome affects connective tissue, which is the “glue” between cells.
- It can affect many parts of the body, such as the skeleton, heart, blood vessels, eyes, skin, nervous system, and lungs.
- The disease is usually passed from parent to child through the genes.
- There is no cure, but treatment can help. Seeing the doctor regularly is important to treat or even prevent some problems.
- Talk to your doctor before starting an exercise program or if you are pregnant or planning to become pregnant.

Marfan syndrome causes problems with connective tissue, which usually holds your body together and provides support for it to grow. Because connective tissue is found throughout the body, Marfan syndrome can affect many parts of the body, including the skeleton, eyes, heart, blood vessels, nervous system, skin, and lungs.

Symptoms

Marfan syndrome affects different people in different ways. Some people have only mild symptoms, while others are more severely affected. In most cases, the symptoms get worse as you get older.

The parts of the body most affected are:

- **Skeleton.** People with Marfan syndrome tend to be tall, slender, and loose-jointed. Other symptoms include:

    - Arms, legs, fingers, and toes that are disproportionately long in relation to the rest of the body.
    - Long, narrow face.
    - Crowded teeth because the roof of the mouth is arched.
    - Protruding or indented breastbone, curvature of the spine, and flat feet.
- **Eyes.** The eyes are commonly affected in Marfan syndrome. Symptoms can include:
    - The lens of one or both eyes is slightly higher or lower than normal, or shifted to one side.
    - Retinal detachment, which is a serious condition.
    - Nearsightedness.
    - Glaucoma (high pressure within the eye).
    - Cataracts (cloudiness of the eye's lens).
- **Heart and blood vessels.** Many people with Marfan syndrome have problems with the heart and blood vessels, including:
    - Weakened or stretched aorta (the large artery that carries blood from the heart to the rest of the body); this increases the risk for serious heart problems or even sudden death.
    - Heart murmur, or leaky heart valves. Small leaks don’t cause problems, but larger ones may cause shortness of breath, tiredness, and a very fast or irregular heart rate.
- **Nervous system.** The dura, or tissue covering the brain and spinal cord, may weaken and stretch in older people with Marfan syndrome. This can cause:
    - Mild discomfort in the back.
    - Pain that spreads to the abdomen.
    - Pain, numbness, or weakness in the legs.
- **Skin.** Issues with the skin include:
    - Stretch marks.
    - Bulge or hernia in the abdomen.
- **Lungs.** Problems with breathing are less common but can include:
    - Lung collapse when tiny air sacs in the lungs become stretched or swollen.
    - Snoring or sleep apnea (brief periods when breathing stops while sleeping).

Causes

Marfan syndrome is caused by a problem with a gene involved in making connective tissue. You were born with the disorder, even though it may not be diagnosed until later in life. Even though everyone with Marfan syndrome has the defective gene, not everyone has the same symptoms.

If you have Marfan syndrome your child has a 50 percent chance of getting the disease. It is rare for two parents without the disease to have a child with it.

Diagnosis

There is no specific test for Marfan syndrome. Your doctor will:

- Ask for information about any family members with the disorder or who had an early, unexplained, heart-related death.
- Give you a physical examination, and measure how long your arms and legs are in proportion to your trunk.
- Examine your eyes.
- Use a special device to test your heart and aorta.
- Perform a genetic analysis, although this can be time consuming and less informative.
- Give you a diagnostic test to rule out a similar disorder, Loeys-Dietz syndrome.

Treatment

There is no cure for Marfan syndrome, although treatment can help. The earlier a potential problem is found, the more likely you can prevent complications. You should see your doctor for regular check-ups to look for changes in the following:

- **Skeleton (spine and sternum).** In some cases, an orthopedic brace or surgery may be recommended.
- **Eyes.** In most cases, eyeglasses or contact lenses can correct eye issues, although surgery may be necessary in some cases.
- **Heart and blood vessels.** If you have heart problems you should wear a medical alert bracelet and go to the emergency room if you have chest, back, or abdominal pain. Treatments can include:
    - Medications to control heart valve problems by decreasing stress on the aorta.
    - Surgery to repair the aorta or replace a valve. Artificial heart valves can increase the risk for blood clots or heart inflammation. Doctors will put you on blood thinners if you have a valve replaced, and your dentist may recommend you take medications prior to any dental work. There is a type of surgery that allows people to keep their own valves.
- **Nervous system.** Medication can help reduce pain if the tissue covering the spinal cord begins to swell.
- **Lungs.** You should not smoke, since you have higher risk for lung damage. You should talk to your doctor about breathing problems when sleeping.

Pregnancy. Women with Marfan syndrome can have healthy babies. However, pregnancy is considered high risk because it stresses the heart. There are some things you can do to reduce this risk:

- Talk to your doctor if you are planning to get pregnant. In some cases, you may need heart valve surgery prior to pregnancy.
- See your doctor on a regular basis to prevent problems with the heart while pregnant.

You might also wish to see a genetic counselor, who can tell you the likelihood of passing the disease to your children.

Who Treats

Doctors who may treat Marfan syndrome include:

- Family doctor or pediatrician, who oversees routine health care.
- Cardiologist, who treats issues with the heart.
- Orthopaedist, who treats bone problems.
- Ophthalmologist, who treats eye disorders.
- Geneticist, who can explain how your genes contributed to your disease andwho can tell you the likelihood that your child will inherit the disease.

Living With

Diagnosis and treatment of Marfan syndrome have improved over the last few decades. These advances make it possible for people with Marfan syndrome to live about as long as the average person.

However, Marfan syndrome can cause strong emotions, such as anger and fear. You may also be worried that you children will have the disease. Some children with Marfan syndrome are told to restrict their activities. This may be hard for a child to understand or accept.

Ways to cope with Marfan syndrome include:

- Receive appropriate medical care and accurate information.
- Obtain social support.
- Eat a balanced diet and maintain a healthy lifestyle.
- Get moderate exercise, which is important to keep the skeleton and heart healthy. Because of possible problems with the aorta, you should not play in contact sports, competitive athletics, or do exercises where you tighten the muscles without moving them. “Planks” are an example of this last type of exercise.
- Receive genetic counseling, which may also be helpful for understanding the disease and determining the likelihood you will pass it on to your children.

Research Progress

Research on Marfan syndrome is exploring genetics, biology, causes of heart complications, and how Marfan syndrome changes the skeleton. Other scientists are focusing on ways to treat

complications associated with Marfan syndrome.

Related Resources

U.S. Food and Drug Administration
Toll free: 888-INFO-FDA (888-463-6332)
Website: https://www.fda.gov

Drugs@FDA at https://www.accessdata.fda.gov/scripts/cder/daf. Drugs@FDA is a searchable catalog of FDA-approved drug products.

Centers for Disease Control and Prevention, National Center for Health Statistics
Website: https://www.cdc.gov/nchs

National Heart, Lung, and Blood Institute
Website: https://www.nhlbi.nih.gov

National Human Genome Research Institute
Website: https://www.genome.gov

Marfan Foundation
http://www.marfan.org

National Organization for Rare Disorders
https://www.rarediseases.org

American Heart Association
https://www.americanheart.org

March of Dimes Birth Defects Foundation
http://www.marchofdimes.com

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at NIAMSInfo@mail.nih.gov.

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